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Letter to the Editor

Narrowing the Map of a Gene (MRXS9) for X-Linked Mental Retardation, Microcephaly, and Variably Short Stature at Xq12-q21.31

To the Editor:

We reported the mapping of a gene (MRXS9) for X-linked mental retardation, microcephaly, and variable short stature to Xq12-q21.31 in 1999 [Shrimpton et al., 1999]. The gene was found to be located between the loci AR and DXS1217, which is about 17 cM over 25 mb in distance.

One member of the original family recently gave birth to another son (IV-3 of Fig. 1), who was born at term weighing 3,418 g, length 51.5 cm, occipitofrontal circumference (OFC) 34 cm (25th centile). At 5 weeks his weight was 4,200 g (25–50th centile), length 52.5 cm (10–25th centile), and OFC 37.5 cm (50th centile). He smiled at around 2 months and rolled over at 5 months. His OFC at 5 months was 43.5 cm (50–75th centile), which was similar to the OFC of his affected older brother at 20 months old. He had surgery for pyloric stenosis at 3 weeks but is otherwise healthy and his psychomotor development is normal. Thus, he seems to be unaffected.

The DNA study showed that he is a recombinant within the previously identified critical region, which allowed us to further narrow the mapping region by excluding a proximal section of the critical region. The new critical region is now between the loci DXS8040 and DXS1217, which is about 11 cM over 22 mb in distance. The LOD score increased from 4.42 to 4.75.

REFERENCE

Shrimpton AE, Daly KM, Hoo JJ. 1999. Mapping of a gene (MRXS9) for X-linked mental retardation, microcephaly, and variably short stature to Xq12-q21.31. Am J Med Genet 84:293–299.

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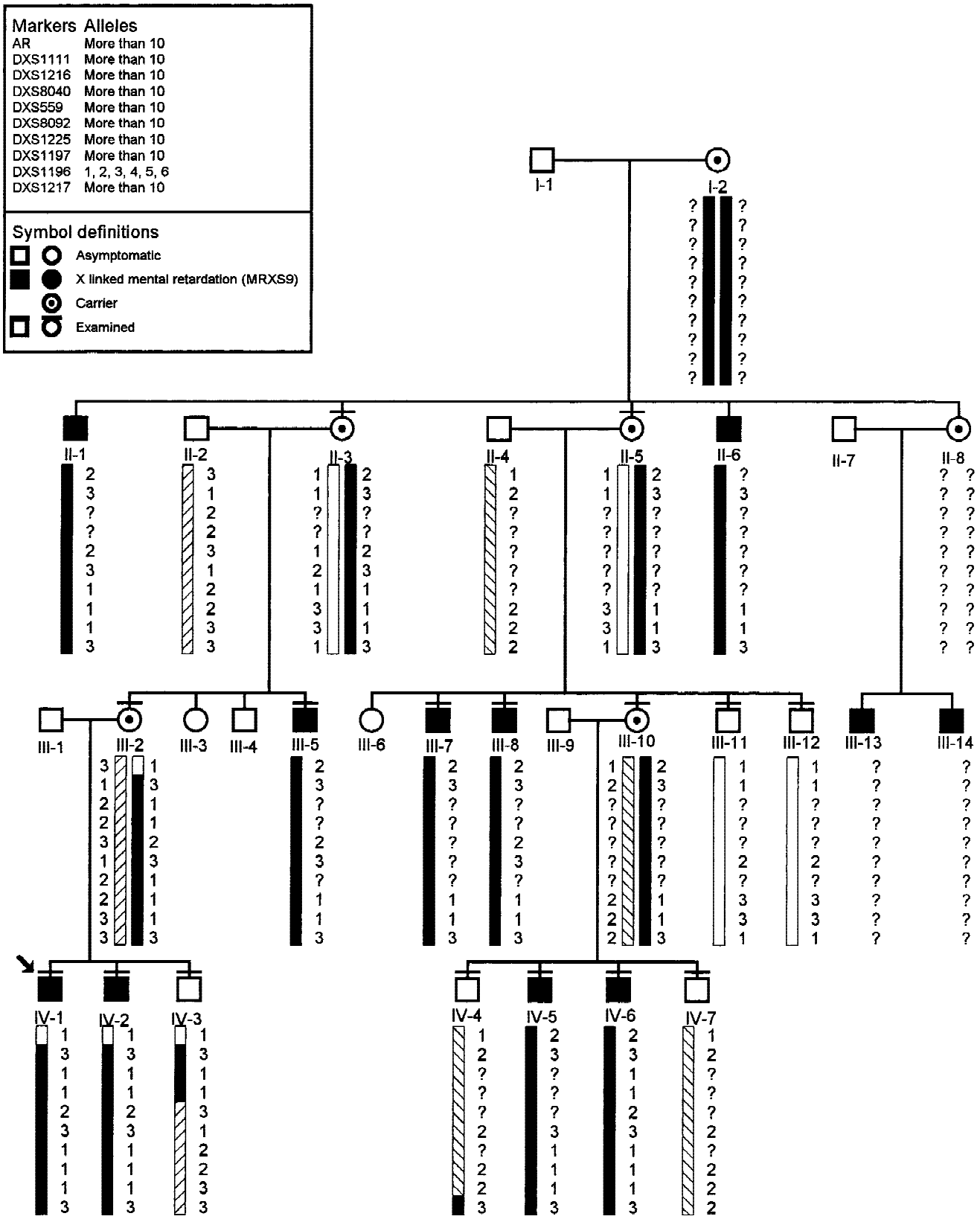

Fig. 1. Revised MRXS9 pedigree showing microsatellite marker haplotypes located on proximal Xq.